

# Correspondence

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## Trisomy 5p syndrome

SIR,

DiLiberti *et al.* (1977) suggested that trisomy 5p has a clinically recognisable and consistent phenotype. In their report on 'Complete trisomy 5p', Brimblecombe *et al.* (1977) suggest that the phenotype in trisomy 5p is non-specific. I have also found three cases (males aged 22, 23, and 7 years) in a family with a partial trisomy 5p13→pter, and each of these patients had the following features; a full face with the appearance of jowls, a long philtrum, convergent strabismus, a bulbous nose, mild malar hypoplasia, prognathism, obesity, and mental retardation. The dermatoglyphs showed a characteristic combination of patterns: an excess of ulnar loops on the fingertips, transversal direction of the dermal ridges on the palms, *t'* triradii, a unilateral, atypical simian crease, and tibial arches on the hallucal areas. A high total finger ridge count was found in only one case (case 3, TFRC = 167; mean value in the normal male population,  $\bar{x}$  = 145, Rodewald *et al.*, 1977; case 1, TFRC = 102; case 2, TFRC = 100).

Several of the dermatoglyphic manifestations in the 5p+ syndrome are strikingly different from features described in cases with partial monosomy 5p-; a high frequency of whorls on the fingertips, longitudinal direction of the dermal ridges on the palms, *t* or *t'* triradii or both, a typical simian crease, and distal loops on the hallucal areas. This contrast confirms, in the context of chromosome 5, the hypothesis of a 'contre type' that Lejeune *et al.* (1964) proposed for the clinical picture of trisomy 21 (Down's syndrome) and its countertype, monosomy 21.

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## Single crease on the 5th finger in medical disorders and in normal population

SIR,

In their recent report on 'Clinical experience with trisomies 18 and 13', Hodes *et al.* (1978) found a single flexion crease on digit 5 in 63% (20 palms in 32 patients) of trisomy 18 patients and in only 5% (1 palm in 19 patients) in trisomy 13 patients. Penrose (1931) first described the presence of a single crease on digit 5 in 16 of 60 cases of Down's syndrome (26.7%). In 8 of these, the single crease was present on one hand only. We found this peculiarity in a group of 200 patients with Down's syndrome (trisomy 21), in 21.5% symmetrically and in 4.5% asymmetrically. The presence of the single crease on both hands (symmetrical) was found in 26.4% of the male patients and only in 13.4% of the female patients. This pattern has also been described in 79.1% of patients with trisomy 18 by Schaumann and Alter (1976), in 43.2% (44 palms) of patients with partial trisomy 9p by Rodewald (1979), and occasionally in partial monosomy 18q (Lejeune *et al.*, 1966), in de Lange's syndrome (Pfeiffer and Kumbhani, 1966; Smith, 1966), and in other medical disorders (Schaumann and Alter, 1976). Cummins and Midlo (1961) noted the absence of the distal interphalangeal flexion crease on an immovable distal joint on the left ring finger of a Negro boy. We found a single crease on the 5th finger in two patients with partial trisomy 9p but not in patients with the trisomy 8 syndrome (Rodewald *et al.*, 1977) or patients with partial trisomy 10p (Rodewald and Stengel-Rutkowski, 1978).

So far, a single crease on the 5th finger in a phenotypically and chromosomally normal subject has not been reported. However, we found a single crease on the 5th right finger, without aplasia or hypoplasia of the middle phalanx and with a movable interphalangeal joint, in a phenotypically and chromosomally normal man (Fig).

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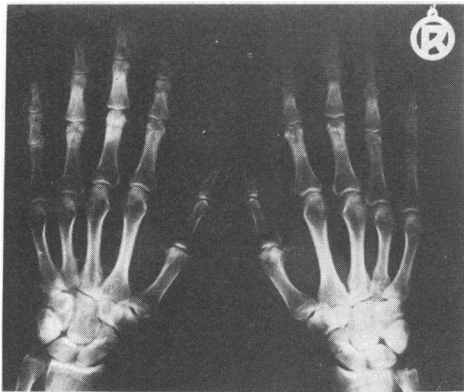


Fig. Print and x-ray of right hand of normal subject.

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creases of the little finger are slightly more common. Komatz *et al.* (1978) found an extra interphalangeal crease in 4 of 551 normal Japanese. An employee who, oddly enough, was responsible for taking dermatoglyphs in our clinic also has this extra interphalangeal crease of the proximal phalanx. Some patients with partial monosomy of 9p have been reported with extra flexion creases, primarily on the second phalanx (DeGrouchy and Turleau, 1977). We recently saw a patient with 'normal' chromosomes and extra flexion creases on the second phalanx of the index fingers and second phalanx of the left ring finger. Because of this finding, the patient was re-examined and was found to have a small deletion of the short arm of chromosome 9. Earlier, another patient with three creases on the right little finger was also found to be partially monosomic for 9p.

Although abnormal flexion creases of the fingers are very strong indicators for chromosomal abnormalities or other syndromes of abnormal development, it appears one cannot say these are never found in normal subjects.

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This letter was shown to Dr Hodes *et al.* who reply as follows:

SIR,

The finding of Rodewald and Wischerath of a normal male with a single flexion crease of the little finger is extremely rare. Extra interphalangeal